SLC17A5 gene

solute carrier family 17 member 5

Normal Function

The *SLC17A5* gene provides instructions for producing a protein called sialin that is located mainly on the membranes of lysosomes, compartments in the cell that digest and recycle materials. Sialin moves a molecule called free sialic acid, which is produced when certain proteins and fats are broken down, out of the lysosomes to other parts of the cell. Free sialic acid means that the sialic acid is not attached (bound) to other molecules.

Researchers believe that sialin may also have other functions in brain cells, in addition to those associated with the lysosomes, but these additional functions are not well understood.

Health Conditions Related to Genetic Changes

sialic acid storage disease

Approximately 20 mutations that cause sialic acid storage disease have been identified in the *SLC17A5* gene. Some of these mutations result in sialin that does not function normally; others prevent sialin from being produced. In a few cases, sialin is produced but not routed properly to the lysosomal membrane.

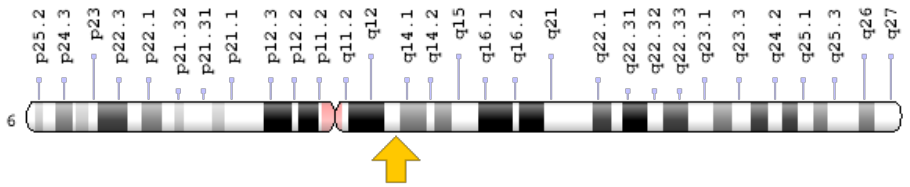
There are three forms of sialic acid storage disease. A particular *SLC17A5* mutation, found primarily in people from Finland and Sweden, causes the least severe form of this disorder known as Salla disease. This mutation replaces the protein building block (amino acid) arginine with the amino acid cysteine at position 39 of the sialin protein (written as Arg39Cys or R39C). Other *SLC17A5* gene mutations that have more damaging effects on sialin protein function cause the most severe form of the disorder, infantile free sialic acid storage disease. Individuals diagnosed with intermediate severe Salla disease have one copy of the *SLC17A5* gene with the Salla disease mutation in each cell, while the second copy of the gene bears a more severe mutation. The severity of signs and symptoms of intermediate severe Salla disease falls between those of Salla disease and infantile free sialic acid storage disease.

SLC17A5 gene mutations that reduce or eliminate sialin activity result in a buildup of free sialic acid in the lysosomes. It is not known how this buildup, or disruption of other possible functions of sialin in the brain, causes the specific signs and symptoms of sialic acid storage disease.

Chromosomal Location

Cytogenetic Location: 6q13, which is the long (q) arm of chromosome 6 at position 13

Molecular Location: base pairs 73,593,378 to 73,654,014 on chromosome 6 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- AST
- ISSD
- NSD
- S17A5_HUMAN
- SD
- SIALIN
- SIASD
- SLD
- solute carrier family 17 (acidic sugar transporter), member 5
- solute carrier family 17, member 5

Additional Information & Resources

GeneReviews

- Free Sialic Acid Storage Disorders
  https://www.ncbi.nlm.nih.gov/books/NBK1470

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28sialic+acid+storage+disease%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D

OMIM

- SOLUTE CARRIER FAMILY 17 (ACIDIC SUGAR TRANSPORTER), MEMBER 5
  http://omim.org/entry/604322

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_SLC17A5.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=SLC17A5%5Bgene%5D
- HGNC Gene Family: Solute carriers
  http://www.genenames.org/cgi-bin/genefamilies/set/752
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=10933
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/26503
- UniProt
  http://www.uniprot.org/uniprot/Q9NRA2

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